International Congress Series 1261 (2004) 350–352

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# Y chromosome STRs in the north of Portugal

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**Abstract.** The interest in Y chromosome polymorphisms has continuously increased, not only in evolutionary/genealogical studies, but also in forensic genetics. The objective of this study is to analyse the *loci* DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385I/II in a population sample from the North of Portugal. One hundred one unique haplotypes were found and the haplotypic diversity was $0.998 \pm 0.001$ which reinforces the forensic utility of these markers. Phylogenetic analysis shows that our population can be included in the European Y chromosome lineages and distinct from the Asian ones. © 2003 Published by Elsevier B.V.

*Keywords:* Y-STR; Minimal haplotype; Population data

## 1. Introduction

The NRY part of the Y human chromosome is paternally inherited and haploid [1] and the interest in Y chromosome polymorphisms has continuously increased. From the forensic point of view, these markers, being passed down without any recombination, can be used in cases of unknown parentage [2]. Moreover, the Y chromosome specific markers are of election in cases of sexual assaults, since they permit the detection of male DNA in mixtures of male/female cells, even with a high background of female DNA, particularly if more than one male is involved [3]. From the evolutionary point of view, these markers are useful for tracing migration movements of populations, inferring the history of modern humans [4]. Having this in mind, we have studied the *loci* DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385I/II in a population sample from the North of Portugal, involved in paternity testing.

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0531-5131/ 
doi:10.1016/S0531-5131(03)01839-9

## 2. Material and methods

DNA was isolated from 122 unrelated healthy individuals, using Chelex or phenol-chloroform extraction methods. Amplification was performed in three PCR reactions: a pentaplex (DYS19, DYS389I, DYS389II, DYS390, DYS393), a triplex (DYS391, DYS392, DYS393) and a monoplex (DYS385I/II) [5,6]. The PCR products were typed by capillary electrophoresis (ABI 310 Genetic Analyser, Applied Biosystems). Haplotypes were determined by simple counting and several statistic parameters were calculated using Arlequin software ver. 2000: gene diversity and variance by AMOVA. Genetic distances were also calculated and the correspondent filogenetic tree obtained as well as multi dimensional scalling (MDS).

## 3. Results and discussion

The analysis of these Y-STR markers showed 111 unique haplotypes. The two most frequent haplotypes were repeated three times and seven others only two times. The highest gene diversity corresponds to DYS385I/II (0.847), followed by DYS389II (0.677), being the DYS389I the locus that presents the lowest value (0.535). The mean gene diversity was 0.626 and the value of the haplotypic diversity was $0.998 \pm 0.001$, probably due to the scarce repetition of haplotypes. So, the probability that two randomly chosen individuals from our population share the same haplotype is 0.002.

Comparisons of our population with others from Iberia, Europe and Asia were performed. We did not compare our haplotypic data with African populations. We share many haplotypes with Iberia: seven with Asturias (North Spain), six with Barcelona (Mediterranean part) and five with Andalusia (South Spain). The number of shared haplotypes decreases progressively with other European populations: four with Germany,

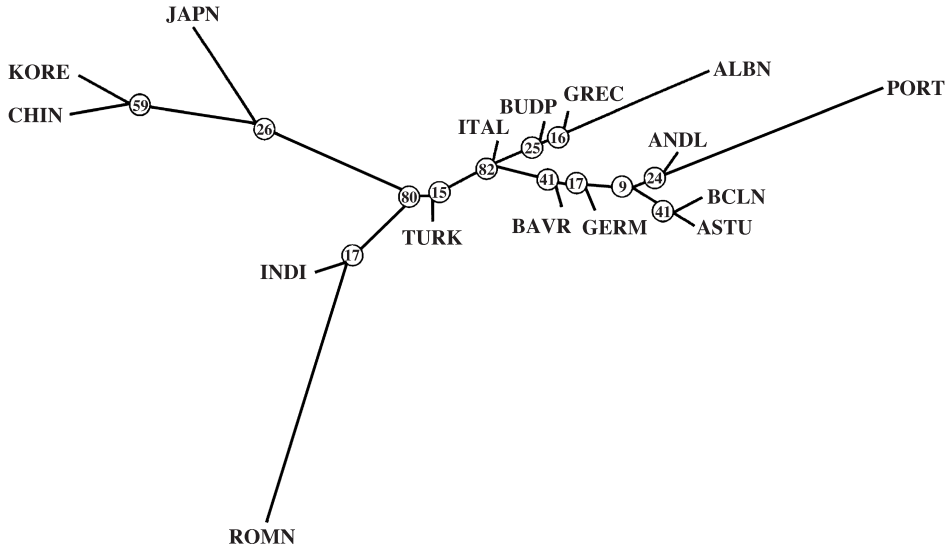


Fig. 1. Neighbour-joining tree based on Slatkin's distances.

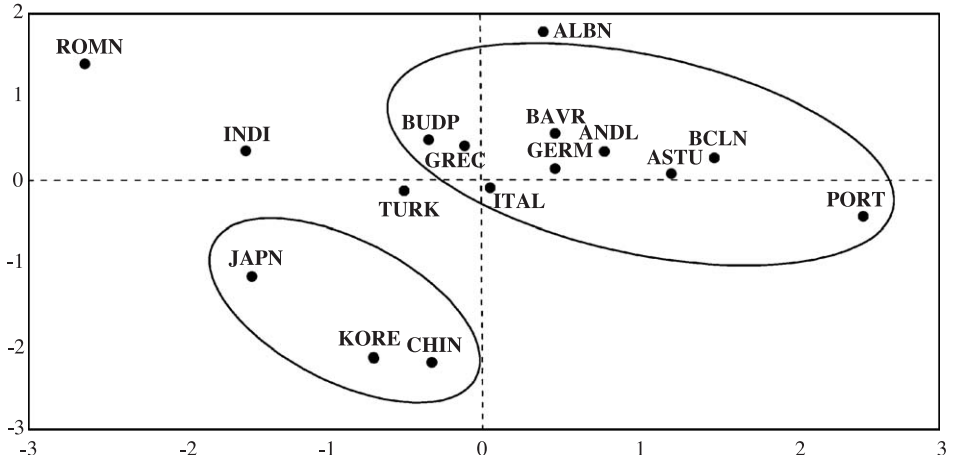


Fig. 2. Multi-dimensional scaling: total variance of 96.4% and the stress coefficient was 0.088.

three with Hungary, two with Italy and one with Turkey, Albania and Romanies. The haplotype 14-13-29-24-11-13-13-11/14 is the most frequent in our population and in all the other European ones compared in this study, with the exception of Italy. None of the haplotypes is shared with India, China, Korea or Japan. The AMOVA analysis shows that the biggest percentage of variance occurs within populations. The Slatkin's genetic distancies [7] between our population and the others were calculated and used to debug a dendrogram (Fig. 1) and a MDS diagram (Fig. 2). The phylogenetic analysis shows that our Y chromosomes can be included in the European lineages and are distinct from the Asian ones. In conclusion, we can reinforce the forensic utility of these markers, namely in sexual assaults. Additionally, the phylogenetic data allows to include the Y chromosomes from the North of Portugal into the European lineages and to distinguish them from the Asian ones.

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